Thalassaemia – A Comparative Analysis Across Europe

a report by
Androulla Eleftheriou

Executive Director, Thalassaemia International Federation, Nicosia

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Thalassaemias were traditionally believed to occur only in regions of the world where malaria is or was endemic, including the Mediterranean, the Middle East, South and East Asia, the Pacific and South China, with carrier rates varying from 2 to 25%. It is now known that haemoglobin disorders occur widely across the world, including in northern and western Europe, not in the indigenous population but as a result of global population movements. Thalassaemias and sickle cell disease are increasingly common in these countries and – with the exception of Spain and Portugal, where the incidence rate is approximately 1–2% – incidence rates are as high as those historically found in southern Europe (4–18%), the region where haemaglobinopathies were first recognised and where successful control programmes were first developed. In southern Europe and especially in the Mediterranean countries, annual affected birth rates have been reduced dramatically (by 80–100%), with Cyprus for example reaching between zero and three affected births each year since 1987, with open-ended survival and very satisfactory quality of life for patients. Today, in these countries the only factors contributing to development of complications, low survival and poor quality of life are related to poor adherence to clinical management. These are the standards the rest of the Europe should aim to reach. In northern and western Europe, in recent years health professionals and policy-makers throughout the region have been confronted with the challenge of setting up, providing and making accessible adequate services to 'at-risk' populations for prevention and treatment of haemoglobin disorders. Accurate epidemiological data, a prerequisite for developing health policies, are to date very limited. A recent study[1] attempted to identify the proportion at risk of haemoglobin disorders in European countries. Data concerning country of birth and ethnic origin of at-risk populations were adjusted to provide estimated prevalence rates. Clearly, accurate information and significant micro-mapping are required to guide the activities and efforts of national health authorities in these countries. This is particularly difficult as ethnic groups are widely scattered throughout Europe.

In northern and western European countries, in contrast to countries with limited resources, national health systems and infrastructures are available for developing national strategies for effective prevention and optimal treatment of haemoglobin disorders. However, groups at increased risk are unevenly distributed and often concentrated in major cities, so establishing equal access to such services is an important issue. The support of national public health authorities and political powers is required to ensure awareness in health professionals and the public and equal access to carrier detection, genetic counselling and pre-natal diagnosis. The UK is a good example of how the absence of a national control programme and lack of sufficient access to the available laboratory and clinical services can lead to ineffective prevention and poor quality of life in patients treated outside expert centres (most of which are found in London). For example, 60% of couples who are at risk in London use pre-natal diagnosis compared with significantly fewer (~20%) living elsewhere in the country. In a study published in 2000 by Modell et al.,[2] 50% of patients with thalassaemia major in the UK died at the age of <35 years, although better survival was seen in expert centres such as University College Hospital, where 40-year-old patients demonstrated 78% survival. In 2004, with the aim of improving this situation, the UK national health authorities launched a new national linked antenatal and neonatal screening programme for sickle cell and thalassaemia. Similar efforts are being made to promote national control policies for haemoglobin disorders in countries throughout western Europe, including Austria, Germany, Belgium, Denmark, The Netherlands, France and Scandinavia. However, considerable work is still needed. The real problem lies in reaching and delivering appropriate services to those at risk and developing effective policies for controlling haemoglobin disorders, although such policies will be more effective if developed with the aim of reaching everyone. For example, France – like the UK – is host to large immigrant populations; although neonatal screening has been available since 1994, effectiveness was lower than expected because it targeted only the so-called at-risk population, which through the years has become integrated into the indigenous population.

Countries in eastern Europe, including Bulgaria, Romania and Albania, are also affected, albeit with carrier rates lower than 3% and, like the Mediterranean and southern European countries, in the context of their indigenous populations. Historically, weak health service infrastructure has hindered epidemiology work and national treatment programmes in these countries. The recent recognition of the burden of these diseases and the general improvement of healthcare systems over the years have allowed progress to be made in the management of haemoglobin disease, but less so in the area of prevention; this is hindered by the lack of accurate epidemiological data. There is a strong case for a pan-European collaboration on haemoglobin disorders in order to enable countries to benefit from each other's experience.[3] Thalassaemia constitutes the first and best monogenic disease, the control of which can be used as a model. Some of the tools that contributed to the successful development of effective control programmes for haemoglobin disorders in southern Europe and that need to be utilised throughout Europe include: development of standards and guidelines for laboratory services; formulation of national guidelines for the management of thalassaemia and the establishment of reference centres; epidemiological studies (micro-mapping for the identification of populations at risk is a prerequisite to the development of appropriate services); establishment of educational programmes for health professionals, patients and the community; and promotion of patient/parental support groups. Other issues that need to be addressed in order to offer holistic services for haemoglobin disorders in these countries include the increasing prevalence of α-thalassaemia, cultural, linguistic and religious differences and the social and genetic implications of consanguinous marriage. ■

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